EUROPEAN JOURNAL OF BIOMEDICAL AND PHARMACEUTICAL SCIENCES
http://www.ejbps.com

ISSN 2349-8870
Volume: 6
Issue: 3
427-428
Year: 2019

MAYER ROKITANSKY KUSTER HAUSER SYNDROME: A RARE CASE REPORT

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Article Received on 10/12/2019 **Article Revised on 01/02/2019** **Article Accepted on 22/02/2019**

INTRODUCTION

Mayer Rokitansky Kuster Hauser Syndrome is characterized by absent uterus and aplasia of upper 2/3rd of vagina with normal karyotyping (46XX) with normal development of secondary sexual characters.[1]

CASE REPORT

A 24 yr old married female came to gynaecology OPD with complaints of lump in abdomen since 10 days. She also had c/o primary amenorrhoea Her marital life was normal and she reported to have no coital complaints

O/E

- The patient's built was normal
- The breast development was tanner stage IV
- Pubic and axillary hair were absent.

On P/A

- 12 X 10cm cyst felt in Rt iliac fossa tenderness present mobile

L/E

- The vagina was 3 inches in length and appeared normal.

On P/S examination cervix could not be visualized
P/V uterus not felt cyst felt in rt iliac fossa mobile tenderness present

The patient had 2 yrs of married life and still could not conceive She got admitted in some outside hospital with complaints of primary amenorrhoea and primary infertility where she had karyotyping which came out to be 46XY and was suggestive of Androgen Insensitivity Syndrome She came to our hospital with this karyotyping report and with complaints of lump in abdomen and others as described above Subsequent investigations were done CT scan revealed complex cystic mass lesion in pelvis extending into the infraumbilical region Further the uterus was not visualized Bladder was displaced inferiorly The testes was not seen (i/v/o Androgen Insensitivity Syndrome) Hence, CT scan was suggestive of complex ovarian cyst with absent testes Her tumor markers were done like HCG , AFP, Ca125 which came out to be normal, Only her AMH was raised Explor Lapratomy was done Intraop findings

- Left sided ovarian cyst was seen of size 12 x 10 cm
- Uterus was not visualized
- Right ovary was seen which appeared bulky
- No testes seen
- Vagina appeared normal
- Left side cystectomy was done

Cyst was sent for Histopathology report which was suggestive of serous cyst adenoma of ovary. As her vagina was normal and adequate in length vaginoplasty was not done. Hence this patient can lead normal sexual life but can't conceive Repeat karyotyping was done which came out to be 46 XX.

DISCUSSION

Mayer – Rokitansky – Kuster – Hauser Syndrome is a congenital disorder which mostly affects the reproductive system of females Primary amenorrhea and lack of sexual development occur in gonadal dysgenesis due to missing ovaries. Primary amenorrhea with sexual development occurs in Rokitansky syndrome due to absence of the uterus, with normal ovarian function. The association of these two conditions has been previously described as a rare event.[2]

Here the uterus and upper 2/3rd of vagina are not developed, although external genitalia are normal This is mostly due to failure of development of Mullerian ducts Mayer-Rokitansky-Kuster-Hauser syn-drome (MRKHS) is a congenital malformation charac-terized by an absence of the vagina associated with a variable abnormality of the uterus and the urinary tract but functional ovaries. The various urinary tract anomalies described are renal agenesis, pelvic kidney, fusion anomaly like horse-shoe kidney and vesicoureteric reflux.

TREATMENT

Uterine transplantation (still in experimental stage) As ovaries are present so this people can have genetic children through IVF Adoptation is also one of the option Vaginoplasty Psychological support is the most important thing for his women.

CONCLUSION

As demonstrated by the present case, this female has a karyotyping of 46 XX with normal vagina with absent uterus with B/L bulky ovaries. Carrier testing should be done within the family as this case has familial tendencies The present case is the only case in her family hence no carrier testing adviced.

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